Patient Information Publications
Warren Grant Magnuson Clinical Center
National Institutes of Health

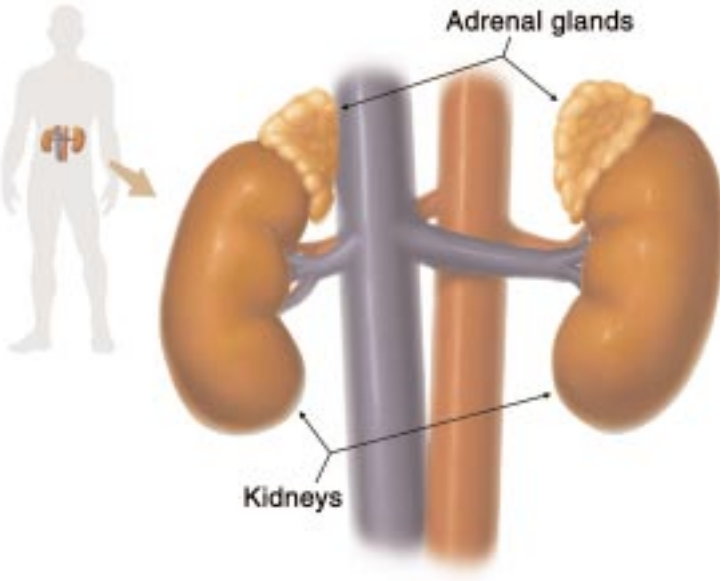
Facts about CAH
Congenital Adrenal Hyperplasia

This information was prepared by your health care team to help you learn about CAH. CAH stands for congenital adrenal hyperplasia. It is a genetic disorder of the adrenal glands that affects the body's general health, growth, and development.

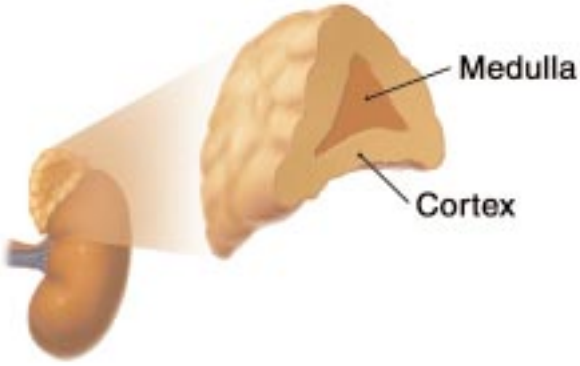
What are the adrenal glands?

The adrenal glands are a pair of walnut-sized organs above the kidneys. They make hormones, which act like chemical messengers to affect other organs in the body.

An organ at the base of the brain, called the pituitary gland, helps regulate the adrenal glands.



Each adrenal gland has two parts: the medulla (the inner part), and the cortex (the outer part). The medulla makes the hormone adrenaline. The cortex makes the hormones cortisol, aldosterone, and androgens. CAH affects how the adrenal cortex works.



What do adrenal hormones do?

Hormones made by the adrenal glands are important for the body's normal function. Cortisol affects energy levels, sugar levels, blood pressure, and the body's response to illness or injury. Aldosterone helps maintain the proper salt level. Androgens are male-like hormones needed for normal growth and development in both boys and girls.

What is CAH?

The adrenal glands help keep the body in balance by making the right amounts of cortisol, aldosterone, and androgens. But in CAH, production of cortisol is blocked. Some children with CAH also lack aldosterone. These imbalances cause the adrenal gland to make too much androgen.

Symptoms
Too little cortisol may cause tiredness and nausea. During illness or injury, low cortisol levels can lead to low blood pressure and even death.

Lack of aldosterone, which occurs in three out of four patients with CAH, upsets salt levels. This imbalance may cause dehydration (too little fluid within the body), and possibly death. Chronic salt imbalance may also cause abnormal growth.

Too much androgen causes abnormal physical development in children. Boys and girls with CAH may grow too fast, develop early pubic hair and acne, and stop growing too soon, causing short stature. Girls exposed to high levels of androgens before birth may have abnormal external genitalia at birth. Although their internal female organs are normal, excess androgens may also affect puberty and cause irregular menstrual periods.

Too much cortisol replacement also causes abnormal development in children. Side effects include obesity and short stature. Also, too much hydrocortisone, the medicine that replacese cortisol in the body, can lower bone density (osteoporosis) and raise cholesterol.

Are there different types of CAH?

There are many types of CAH. The most common is 21-hydroxylase deficiency (95 percent of cases). A child with this type of CAH has adrenal glands that cannot make cortisol and may or may not make aldosterone. As a result, the glands overwork trying to make these hormones and end up making too much of what they can make: androgens.

The second most common form of CAH is 11-hydroxylase deficiency. A child with this type of CAH has adrenal glands that make too much androgen and no cortisol. Children with this type of CAH may also have high blood pressure.

Rare types of CAH include 3-beta-hydroxysteroid dehydrogenase deficiency, lipoid CAH, and 17-hydroxylase deficiency.

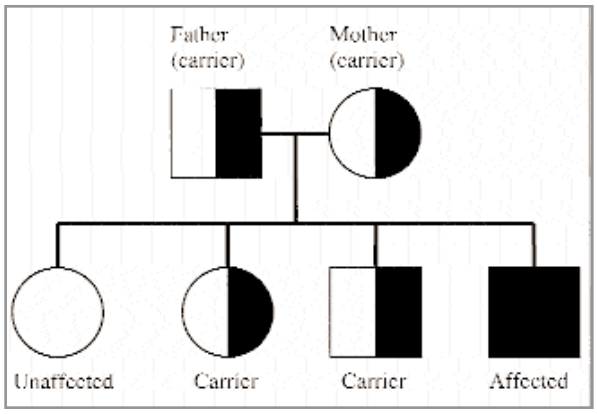
Late-onset (nonclassical) CAH
This type of CAH is a milder form of severe CAH. People with late-onset 21-hydroxylase deficiency make enough cortisol and aldosterone, but they make excess androgens. Symptoms come and go, beginning at any time but typically in childhood or early adulthood. Boys usually do not need treatment.

Girls usually need treatment to suppress their androgens.

Nonclassical CAH is common. One in every 1,000 people has nonclassical 21-hydroxylase deficiency. Incidence is higher in certain ethnic groups: 1 in 27 Ashkenazic Jews, 1 in 53 Hispanics, 1 in 63 Yugoslavs, and 1 in 333 Italians.

How is CAH inherited?

An inherited disorder is one that can be passed from the parents to their children. CAH is a type of inherited disorder called "autosomal recessive."



For a child to have CAH, each parent must either have CAH or carry a genetic mutation. This means that if two parents are CAH carriers (that is, they have the gene for CAH but not the disorder), their children have a 25 percent chance of being born with CAH. Each sibling without CAH has two chances in three of being a carrier. Tests can be done to find out if someone is a carrier.

Classical CAH occurs in 1 in 14,000 births.

How is CAH treated?

The standard treatment for 21-hydroxylase deficiency is hydrocortisone which replaces cortisol, and fludrocortisone (Florinef) which replaces aldosterone.

For 11-hydroxylase deficiency, the treatment is only hydrocortisone.

Because replacement medications cannot mimic the body's exact needs, patients, on average, are about 4 inches shorter than their peers.

What if a child with CAH has an illness, surgery, or a major injury?

During these times, a child with CAH needs closer medical attention and should be under a doctor's care. More cortisol is needed to meet the body's increased needs for this hormone. Higher doses of hydrocorisone are given by mouth or sometimes by injection. Intravenous medication is needed before surgery.

How long can people live with CAH?

People with CAH have a normal life span.

Can a woman with CAH become pregnant and have a baby?

Increased androgens may cause irregular menstrual periods and make it harder for a woman with CAH to conceive a child. But if she takes her medications as directed, she can become pregnant and have a baby.

Do men with CAH have fertility problems?

Men who take medications as directed usually have normal fertility. Rarely, however, they may develop "adrenal rest tissue" in their testicles. This is when adrenal tissue grows in other parts of the body such as the testicles or scrotum. Having adrenal rest tissue may affect a man's ability to father a child. The tissue does not turn to cancer,

but it can grow enough to cause discomfort or infertility. Large growths are rare, and surgery is usually not needed.

Do children with CAH outgrow it?

CAH cannot be outgrown, and treatment is required for life. Treatment is tailored for each child and adjusted during his or her growth.

Can CAH be diagnosed prenatally?

CAH can be diagnosed before birth. Amniocentesis or chorionic villus sampling during pregnancy can check for the disorder.

Prenatal treatment

Experimental prenatal treatment is available. For this treatment, mothers take dexamethasone, a potent form of hydrocortisone. This drug suppresses androgens in the fetus and allows female genitalia to grow more normally. This treatment lessens or eliminates the need for surgery in girls. It does not, however, treat other aspects of the disorder. Children with CAH still need to take hydrocortisone and Florinef for life.

What research is being done?

Researchers are working on many aspects of CAH including discovering new ways to diagnose and treat the disorder and finding the precise genetic defects that cause CAH.

At NIH, scientists are learning more about CAH. They also search for better treatment choices to help children with classical CAH grow and develop.

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